



## SMC1A gene

structural maintenance of chromosomes 1A

### Normal Function

The *SMC1A* gene provides instructions for making a protein that is part of the structural maintenance of chromosomes (SMC) family. Within the nucleus, SMC proteins help regulate the structure and organization of chromosomes.

The protein produced from the *SMC1A* gene (which is usually called the SMC1 protein) helps control chromosomes during cell division. Before cells divide, they must copy all of their chromosomes. The copied DNA from each chromosome is arranged into two identical structures, called sister chromatids, which are attached to one another during the early stages of cell division. The SMC1 protein is part of a protein group called the cohesin complex that holds the sister chromatids together.

Researchers believe that the SMC1 protein, as a structural component of the cohesin complex, also plays important roles in stabilizing cells' genetic information, repairing damaged DNA, and regulating the activity of certain genes that are essential for normal development.

### Health Conditions Related to Genetic Changes

#### Cornelia de Lange syndrome

More than 35 mutations in the *SMC1A* gene have been identified in people with Cornelia de Lange syndrome, a developmental disorder that affects many parts of the body. Researchers estimate that mutations in this gene account for about 5 percent of all cases of this condition.

Most of the *SMC1A* gene mutations that cause Cornelia de Lange syndrome change single protein building blocks (amino acids) in the SMC1 protein. These mutations alter the structure and function of the protein, which likely interferes with the activity of the cohesin complex and impairs its ability to regulate genes that are critical for normal development. Although researchers do not fully understand how these changes cause Cornelia de Lange syndrome, they suspect that altered gene regulation probably underlies many of the developmental problems characteristic of the condition.

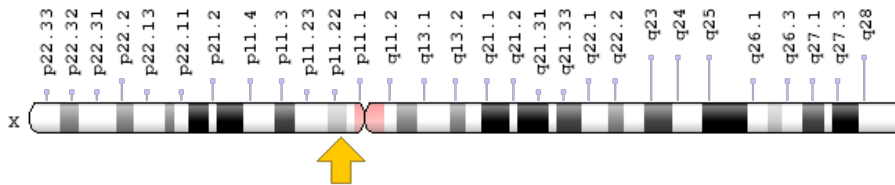
Studies suggest that mutations in the *SMC1A* gene tend to cause a form of Cornelia de Lange syndrome with relatively mild features. Compared to mutations in the *NIPBL* gene, which are the most common known cause of the disorder, *SMC1A* gene

mutations often cause less significant delays in development and growth and are less likely to cause major birth defects.

### Chromosomal Location

Cytogenetic Location: Xp11.22, which is the short (p) arm of the X chromosome at position 11.22

Molecular Location: base pairs 53,374,149 to 53,422,728 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- DXS423E
- KIAA0178
- segregation of mitotic chromosomes 1
- SMC1
- SMC1-alpha
- SMC1A\_HUMAN
- SMC1L1
- SMCB

### Additional Information & Resources

#### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Cohesins and Condensins Help Configure Replicated Chromosomes for Segregation  
<https://www.ncbi.nlm.nih.gov/books/NBK26931/#A3334>

#### GeneReviews

- Cornelia de Lange Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1104>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SMC1A%5BTIAB%5D%29+OR+%28SMC1L1%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>

### OMIM

- STRUCTURAL MAINTENANCE OF CHROMOSOMES 1A  
<http://omim.org/entry/300040>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SMC1A.html](http://atlasgeneticsoncology.org/Genes/GC_SMC1A.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SMC1A%5Bgene%5D>
- HGNC Gene Family: Cohesin complex  
<http://www.genenames.org/cgi-bin/genefamilies/set/1060>
- HGNC Gene Family: Structural maintenance of chromosomes proteins  
<http://www.genenames.org/cgi-bin/genefamilies/set/761>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=11111](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11111)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/8243>
- UniProt  
<http://www.uniprot.org/uniprot/Q14683>

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